chromosome 2

Humans normally have 46 chromosomes in each cell, divided into 23 pairs. Two copies of chromosome 2, one copy inherited from each parent, form one of the pairs. Chromosome 2 is the second largest human chromosome, spanning about 243 million building blocks of DNA (base pairs) and representing almost 8 percent of the total DNA in cells.

Identifying genes on each chromosome is an active area of genetic research. Because researchers use different approaches to predict the number of genes on each chromosome, the estimated number of genes varies. Chromosome 2 likely contains 1,300 to 1,400 genes that provide instructions for making proteins. These proteins perform a variety of different roles in the body.

Health Conditions Related to Chromosomal Changes

The following chromosomal conditions are associated with changes in the structure or number of copies of chromosome 2.

2q37 deletion syndrome

2q37 deletion syndrome is caused by a deletion of genetic material from a specific region in the long (q) arm of chromosome 2. The deletion occurs near the end of the chromosome at a location designated 2q37. The size of the deletion varies among affected individuals. The signs and symptoms of this disorder, which may include intellectual disability, autism, short stature, obesity, and characteristic facial features, are probably related to the loss of multiple genes in this region.

cancers

Changes in chromosome 2 have been identified in several types of cancer. These genetic changes are somatic, which means they are acquired during a person's lifetime and are present only in cells that give rise to the cancer. For example, a rearrangement (translocation) of genetic material between chromosomes 2 and 3 has been associated with cancers of a certain type of blood cell originating in the bone marrow (myeloid malignancies).

Trisomy 2, in which cells have three copies of chromosome 2 instead of the usual two copies, has been found in myelodysplastic syndrome. This disease affects the blood and bone marrow. People with myelodysplastic syndrome have a low number of red blood cells (anemia) and an increased risk of developing a form of blood cancer known as acute myeloid leukemia.

SATB2-associated syndrome

Genetic changes on the long (q) arm of chromosome 2 have been found to cause *SATB2*-associated syndrome. Individuals with this condition have intellectual disability and severe speech problems. They may also have an opening in the roof of the mouth (cleft palate), dental abnormalities, or other abnormalities of the head and face (craniofacial anomalies).

Several types of genetic changes are involved in *SATB2*-associated syndrome, all of which affect a gene on chromosome 2 called *SATB2*. Some mutations remove genetic material from the long arm of chromosome 2. These deletions occur in regions designated 2q32 and 2q33, and the size of the deletion varies among affected individuals. They may be large, removing several genes from chromosome 2, including *SATB2*. Or they may be smaller, removing material from within the *SATB2* gene. Other mutations, such as those that change single DNA building blocks (nucleotides) in the *SATB2* gene, can also cause *SATB2*-associated syndrome.

These genetic changes disrupt the *SATB2* gene and are thought to reduce the amount of functional protein produced from it. The SATB2 protein directs development of the brain and craniofacial structures, and a reduction in this protein's function impairs their normal development, leading to the features of the condition.

The signs and symptoms of *SATB2*-associated syndrome are usually similar, regardless of the type of mutation that causes it. However, some individuals with large deletions have uncommon features of the condition, such as problems with the heart, genitals and urinary tract (genitourinary tract), skin, or hair. These features are thought to be related to loss of other genes near *SATB2* on the long arm of chromosome 2.

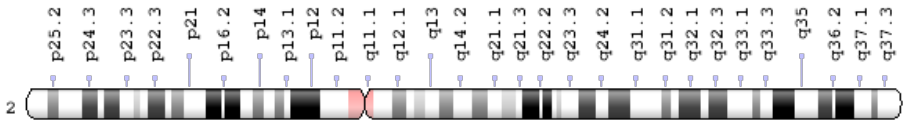
other chromosomal conditions

Another chromosome 2 abnormality is known as a ring chromosome 2. A ring chromosome is formed when breaks occur at both ends of the chromosome and the broken ends join together to form a circular structure. Individuals with this chromosome abnormality often have developmental delay, small head size (microcephaly), slow growth before and after birth, heart defects, and distinctive facial features. The severity of symptoms typically depends on how many and which types of cells contain the ring chromosome 2.

Other changes involving the number or structure of chromosome 2 include an extra piece of the chromosome in each cell (partial trisomy 2) or a missing segment of the chromosome in each cell (partial monosomy 2). These changes can have a variety of effects on health and development, including intellectual disability, slow growth, characteristic facial features, weak muscle tone (hypotonia), and abnormalities of the fingers and toes.

Chromosome Diagram

Geneticists use diagrams called idiograms as a standard representation for chromosomes. Idiograms show a chromosome's relative size and its banding pattern, which is the characteristic pattern of dark and light bands that appears when a chromosome is stained with a chemical solution and then viewed under a microscope. These bands are used to describe the location of genes on each chromosome.



Credit: Genome Decoration Page/NCBI

Additional Information & Resources

MedlinePlus

- Encyclopedia: Chromosome
  https://medlineplus.gov/ency/article/002327.htm

Additional NIH Resources

- National Human Genome Research Institute: Chromosome Abnormalities
  https://www.genome.gov/11508982/

Educational Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology: Trisomy 2
  http://atlasgeneticsoncology.org/Anomalies/Tri2ID1429.html

GeneReviews

- 2q37 Microdeletion Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1158

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Chromosomes,+Human,+Pair+2%5BMAJR%5D%29+AND+%28Chromosome+2%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

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